



## BRCA1 gene

BRCA1, DNA repair associated

### Normal Function

The *BRCA1* gene provides instructions for making a protein that acts as a tumor suppressor. Tumor suppressor proteins help prevent cells from growing and dividing too rapidly or in an uncontrolled way.

The BRCA1 protein is involved in repairing damaged DNA. In the nucleus of many types of normal cells, the BRCA1 protein interacts with several other proteins to mend breaks in DNA. These breaks can be caused by natural and medical radiation or other environmental exposures, and they also occur when chromosomes exchange genetic material in preparation for cell division. By helping to repair DNA, the BRCA1 protein plays a critical role in maintaining the stability of a cell's genetic information.

Research suggests that the BRCA1 protein also regulates the activity of other genes and plays an essential role in embryonic development. To carry out these functions, the BRCA1 protein interacts with many other proteins, including other tumor suppressors and proteins that regulate cell division.

### Health Conditions Related to Genetic Changes

#### breast cancer

Researchers have identified more than 1,800 mutations in the *BRCA1* gene. Many of these mutations are associated with an increased risk of breast cancer in both men and women, as well as several other types of cancer. These mutations are present in every cell in the body and can be passed from one generation to the next. As a result, they are associated with cancers that cluster in families. However, not everyone who inherits a mutation in the *BRCA1* gene will develop cancer. Other genetic, environmental, and lifestyle factors also contribute to a person's cancer risk.

Most *BRCA1* gene mutations lead to the production of an abnormally short version of the BRCA1 protein or prevent any protein from being made from one copy of the gene. As a result, less of this protein is available to help repair damaged DNA or fix mutations that occur in other genes. As these defects accumulate, they can trigger cells to grow and divide uncontrollably to form a tumor.

#### ovarian cancer

Many of the same *BRCA1* gene mutations that increase the risk of breast cancer (described above) also increase the risk of ovarian cancer. Families with these

mutations are often said to be affected by hereditary breast and ovarian cancer syndrome. Women with *BRCA1* gene mutations have a 35 to 60 percent chance of developing ovarian cancer in their lifetimes, as compared with 1.6 percent in the general population.

### prostate cancer

At least five inherited *BRCA1* gene mutations have been found to increase the risk of prostate cancer. These mutations likely reduce the BRCA1 protein's ability to repair DNA, allowing potentially damaging mutations to persist in various other genes. The accumulation of damaging mutations can lead to the out-of-control cell growth and division that can cause a tumor to develop. Men who carry a *BRCA1* gene mutation that increases the risk of prostate cancer may also be at increased risk for other cancers.

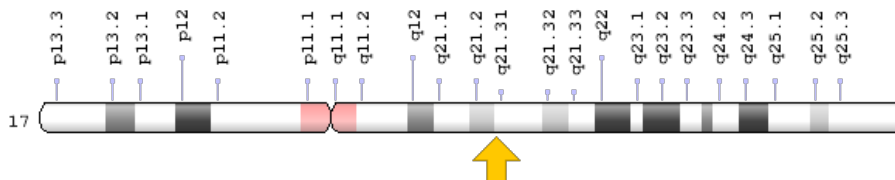
### other cancers

Inherited mutations in the *BRCA1* gene also increase the risk of several other types of cancer, including pancreatic cancer and colon cancer. These mutations impair the ability of the BRCA1 protein to help repair damaged DNA. As defects accumulate in DNA, they can trigger cells to grow and divide without order to form a tumor. It is not clear why different individuals with *BRCA1* mutations develop cancers in different organs. Environmental factors that affect specific organs may contribute to the development of cancers at particular sites.

## **Chromosomal Location**

Cytogenetic Location: 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31

Molecular Location: base pairs 43,044,295 to 43,125,483 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- BRCA1 gene
- BRCA1\_HUMAN
- BRCC1
- breast cancer 1
- breast cancer 1 gene
- breast cancer 1, early onset
- breast cancer 1, early onset gene
- breast cancer type 1 susceptibility gene
- breast cancer type 1 susceptibility protein
- IRIS
- PPP1R53
- PSCP
- RNF53

## Additional Information & Resources

### Educational Resources

- Cancer Medicine (sixth edition, 2003): BRCA1 and BRCA2: Gene Structure and Function  
<https://www.ncbi.nlm.nih.gov/books/NBK12959/#A4564>
- Madame Curie Bioscience Database: Origin, Recognition, Signaling and Repair of DNA Double-Strand Breaks in Mammalian Cells  
<https://www.ncbi.nlm.nih.gov/books/NBK6555/>
- National Cancer Institute: Genetics of Breast and Gynecologic Cancers (PDQ)  
<https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq>

### GeneReviews

- BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer  
<https://www.ncbi.nlm.nih.gov/books/NBK1247>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28BRCA1%5BMAJR%5D%29+AND+%28BRCA1%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

## OMIM

- BREAST CANCER 1 GENE  
<http://omim.org/entry/113705>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/BRCA1ID163ch17q21.html>
- Cancer Genetics Web: BRCA1  
<http://www.cancerindex.org/geneweb/BRCA1.htm>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=BRCA1%5Bgene%5D>
- HGNC Gene Family: BRCA1 A complex  
<http://www.genenames.org/cgi-bin/genefamilies/set/1328>
- HGNC Gene Family: BRCA1 B complex  
<http://www.genenames.org/cgi-bin/genefamilies/set/1335>
- HGNC Gene Family: BRCA1 C complex  
<http://www.genenames.org/cgi-bin/genefamilies/set/1336>
- HGNC Gene Family: Fanconi anemia complementation groups  
<http://www.genenames.org/cgi-bin/genefamilies/set/548>
- HGNC Gene Family: Protein phosphatase 1 regulatory subunits  
<http://www.genenames.org/cgi-bin/genefamilies/set/694>
- HGNC Gene Family: Ring finger proteins  
<http://www.genenames.org/cgi-bin/genefamilies/set/58>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=1100](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1100)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/672>
- UniProt  
<http://www.uniprot.org/uniprot/P38398>

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